



## SMN1 gene

survival of motor neuron 1, telomeric

### Normal Function

The *SMN1* gene provides instructions for making the survival motor neuron (SMN) protein. The SMN protein is found throughout the body, with high levels in the spinal cord. This protein is particularly important for the maintenance of specialized nerve cells called motor neurons, which are located in the spinal cord and the part of the brain that is connected to the spinal cord (the brainstem). Motor neurons control muscle movement.

In cells, the SMN protein plays an important role in processing molecules called messenger RNA (mRNA), which serve as genetic blueprints for making proteins. Messenger RNA begins as a rough draft (pre-mRNA) and goes through several processing steps to become a final, mature form. The SMN protein helps to assemble the cellular machinery needed to process pre-mRNA. Research findings indicate that the SMN protein is also important for the development of specialized outgrowths from nerve cells called dendrites and axons. Dendrites and axons are required for the transmission of impulses between nerves and from nerves to muscles.

A small amount of SMN protein is produced from a gene similar to *SMN1* called *SMN2*. The *SMN2* gene provides instructions for making several versions of the SMN protein, but only one version is functional; the other versions are smaller and easily broken down.

### Health Conditions Related to Genetic Changes

[amyotrophic lateral sclerosis](#)

[spinal muscular atrophy](#)

About 95 percent of individuals with spinal muscular atrophy have mutations that delete a section called exon 7 in both copies of the *SMN1* gene in each cell. As a result, little or no SMN protein is made. In about 5 percent of people with this disorder, one copy of the *SMN1* gene has a deletion of exon 7, and the other copy has a different mutation that disrupts the production or function of the SMN protein. Researchers have identified at least 65 mutations in the *SMN1* gene that cause spinal muscular atrophy.

Motor neurons seem to be particularly vulnerable to a shortage of the SMN protein and die prematurely. Researchers suggest that a shortage of SMN protein leads to

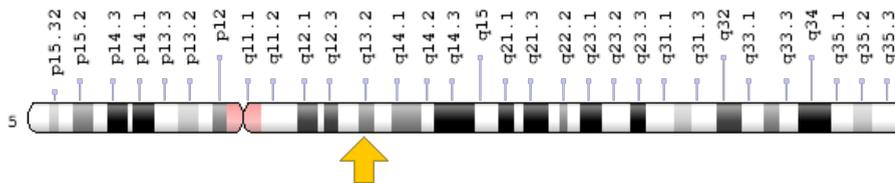
the inefficient assembly of the machinery needed to process pre-mRNA. Without mature mRNA, the production of proteins necessary for cell growth and function is disrupted. Some research findings indicate that a shortage of SMN protein impairs the formation and function of axons and dendrites, possibly leading to the death of neurons. While the cause of neuronal death is unclear, it is the loss of motor neurons that leads to the signs and symptoms of spinal muscular atrophy.

In some cases of spinal muscular atrophy, in addition to their *SMN1* gene mutations, affected individuals have three or more copies of the *SMN2* gene in each cell. Extra *SMN2* genes can help replace some of the SMN protein that is lost due to mutations in the *SMN1* genes. In general, symptoms are less severe and begin later in life in affected individuals with three or more copies of the *SMN2* gene compared with those who have two copies of the gene.

### Chromosomal Location

Cytogenetic Location: 5q13.2, which is the long (q) arm of chromosome 5 at position 13.2

Molecular Location: base pairs 70,924,941 to 70,953,015 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

### Other Names for This Gene

- BCD541
- SMA1
- SMA2
- SMA3
- SMA4
- SMN\_HUMAN
- SMNT

- T-BCD541
- telomeric SMN

## **Additional Information & Resources**

### Educational Resources

- Madame Curie Bioscience Collection: Proteins that Help with the Formation of RNA-Protein Complexes  
<https://www.ncbi.nlm.nih.gov/books/NBK6016/#A43536>
- Molecular Biology of the Cell (fourth edition, 2002): The Nucleus Contains a Variety of Subnuclear Structures  
<https://www.ncbi.nlm.nih.gov/books/NBK26887/#A1048>
- Washington University, St. Louis Neuromuscular Disease Center  
<http://neuromuscular.wustl.edu/synmot.html#smnp>

### GeneReviews

- Amyotrophic Lateral Sclerosis Overview  
<https://www.ncbi.nlm.nih.gov/books/NBK1450>
- Spinal Muscular Atrophy  
<https://www.ncbi.nlm.nih.gov/books/NBK1352>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SMN1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>

### OMIM

- SURVIVAL OF MOTOR NEURON 1  
<http://omim.org/entry/600354>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_SMN1.html](http://atlasgeneticsoncology.org/Genes/GC_SMN1.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=SMN1%5Bgene%5D>
- HGNC Gene Family: Tudor domain containing  
<http://www.genenames.org/cgi-bin/genefamilies/set/780>

- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=11117](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11117)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/6606>
- UniProt  
<http://www.uniprot.org/uniprot/Q16637>

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<https://ghr.nlm.nih.gov/gene/SMN1>

Reviewed: August 2012  
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications  
U.S. National Library of Medicine  
National Institutes of Health  
Department of Health & Human Services